TPO gene

thyroid peroxidase

Normal Function

The *TPO* gene provides instructions for making an enzyme called thyroid peroxidase. This enzyme plays a central role in the function of the thyroid gland, a butterfly-shaped tissue in the lower neck. Thyroid peroxidase assists the chemical reaction that adds iodine to a protein called thyroglobulin, a critical step in generating thyroid hormones. Thyroid hormones play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism).

To function properly, thyroid peroxidase must be located in the cell membrane of certain thyroid cells, called follicular cells. Thyroid peroxidase has several different versions (isoforms), which vary by size and location within the cell. Some versions do not function because they are not located in the cell membrane.

Health Conditions Related to Genetic Changes

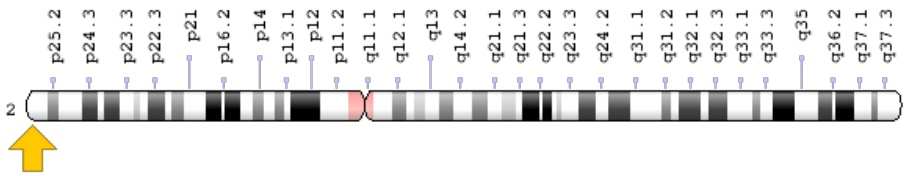
congenital hypothyroidism

TPO gene mutations can cause congenital hypothyroidism, a condition characterized by abnormally low levels of thyroid hormones starting from birth. The *TPO* gene mutations involved in this condition delete, add, or change DNA building blocks (base pairs) in the *TPO* gene. Some mutations lead to an abnormally small thyroid peroxidase enzyme that breaks apart before it can be inserted into the cell membrane. Other mutations change the enzyme's 3-dimensional shape, preventing it from functioning properly within the cell membrane. Without functional thyroid peroxidase, iodine taken up by the thyroid gland is not added to thyroglobulin. As a result, the production of thyroid hormones is absent or reduced, leading to the features of congenital hypothyroidism. In most affected individuals, the thyroid gland is enlarged (goiter) in an attempt to compensate for reduced hormone production. Because cases caused by *TPO* gene mutations result from a disruption of thyroid hormone synthesis, they are classified as thyroid dyshormonogenesis.

Chromosomal Location

Cytogenetic Location: 2p25.3, which is the short (p) arm of chromosome 2 at position 25.3

Molecular Location: base pairs 1,413,461 to 1,542,729 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MSA
- PERT_HUMAN
- thyroid microsomal antigen
- thyroperoxidase
- TPX

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TPO%5BTIAB%5D%29+OR+%28thyroid+peroxidase%5BTIAB%5D%29%29+AND+%28iodide+peroxidase%5BMAJR%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- THYROID PEROXIDASE
  http://omim.org/entry/606765

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_TPO.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TPO%5Bgene%5D
- HGNC Gene Family: Sushi domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1179
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12015
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7173
- UniProt
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